

TITLE OF THE ABSTRACT: A MORPHOLOGICAL STUDY OF ACUTE MYELOID LEUKEMIA AND CORRELATION WITH CLINICAL AND LABORATORY FINDINGS, INCLUDING RESULTS OF CYTOGENETIC ANALYSIS AND MUTATION SCREENING.

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### **Objectives:**

- To **describe the morphology** of bone marrow aspirates **in acute myeloid leukemia(AML)**.
- To **correlate** the bone marrow morphology **with results of cytogenetic analysis and mutation screening**.
- To **categorize AML** using both the French-American-British(**FAB**) and the World Health Organization(**WHO**) **classification** systems

### **Methods:**

- A total of **156 newly diagnosed cases** of AML presenting to Christian Medical College, Vellore over a period of **one year** from January 2015 to December 2015 were included in this study.
- Bone marrow aspirate smears and peripheral blood smears of 156 patients with AML were studied and findings such as differential count, blast lineage assessment and assessment of dysplasia were noted.
- Laboratory haematological parameters such as haemoglobin, platelet count and total leucocyte count were assessed for all the patients.
- Cytogenetic analysis using karyotyping was carried out in **142** of the 156 patients.
- Mutation screening for NPM1, FLT3-ITD and FLT3-TKD genes was carried out for **47** patients.

### **Results**

- The **median age** of the patients was **32 years** (range: 7 months to 76 years).
- Using the **FAB classification**, the most common subtype was **AML-M2(37%)** followed by AML-M1(21%). AML-M6(1%) was the least common subtype.
- **Cytogenetic analysis –**
  - **Abnormal karyotypes** were seen **62%** of our patients.
  - **(15;17)(q22;q12)** was the most common recurrent genetic abnormality observed. t(8;21)(q22;q22.1) was seen in 8% patient. Rest of the abnormalities were seen in ~1-2% of patients.
  - Other common cytogenetic abnormalities observed were
    - Trisomy 8 – 9%
    - Monosomy 7 – 8%

- Complex karyotypes (3 or more abnormalities) – 13%
  - Monosomal karyotypes – 9%.
- **Mutation screening –**
  - **NPM1 mutation** was seen in **47% (22/47)** patients, and in 74% (20/27) patients with normal karyotype.
  - FLT3-ITD mutations were seen in 19% (9/47) of patients.
  - FLT3-TKD mutations were seen in 17% (8/47) of patients.
  - Concomitant NPM1 and FLT3 mutations were concomitantly seen in 28% (13/47) of patients
- Using the **2016 revision to the WHO classification of AML–**
  - **AML with recurrent genetic abnormalities (AML-RGA)** accounted for **42%** of patients.
  - **AML, not otherwise specified (AML, NOS)** accounted for **37%** of patients.
  - **AML with myelodysplasia-related changes(AML-MRC)** accounted for **21%** of patients
    - AML-MRC based only on morphological evidence of dysplasia - 23%.
    - AML-MRC based on morphology and /or cytogenetic changes - 77%.
- **Risk classification** according to modified European LeukemiaNet(ELN) classification system –
  - **Favorable** risk profile was seen in **37%** of patients.
  - **Intermediate** risk profile was seen in **38%** of patients
  - **Adverse** risk profile was seen in **25%** of patients.

**Key words:** Acute myeloid leukemia, AML, FAB classification, WHO 2016 classification, cytogenetics, NPM1, FLT3-ITD, FLT3-TKD, ELN classification.